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Expanding the Role of the Genetic Counselor

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The basic components of genetic counseling are informational and educational. The patient's cognitive and emotional presentation and the needs and concerns of the patient are seldom addressed. Females who carry the FMR1 pre and full gene mutation may present with learning, cognitive, and/or emotional difficulties and family members of those diagnosed with fragile X syndrome have ongoing needs and concerns. As a result, genetic counseling for fragile X syndrome offers a unique opportunity within which to expand the role of the genetic counselor.

Q-methodology, by using the q-sort, is centered on the family to reproduce the needs and concerns that are consistent with the patient's own experience. Used for sociological research, the q-methodology with specially constructed q-sort items is easily adapted to the genetic counseling setting© 1995 Flynn & Gane and can be used for directly assessing the patient's needs and concerns. For our pilot study, 16 items were physically sorted and ranked interdependently by the patient (subject).

Thirty-seven patients (29 females and 8 males) participated in our pilot study. Preliminary results show that the age of the proband, length of time of the diagnosis, and parental sex at the time the q-sort is administered impacts the ranking of items thus differentiating needs and concerns. Results have shown that specific items are missing from the lives of subjects.

From the information obtained from the q-sort, the genetic counselor can identify needs and concerns of the patient and combine this information with the clinical presentation to work with the patient in a more effective manner.

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INTRODUCTION

Since the FMR1 gene was cloned and sequenced [Verkerk et al., 1991; Fu et al., 1991], the number of patients diagnosed with the pre and full gene mutation has increased [Rousseau et al., 1994]. This development has led to an increasing number of family members being impacted by the diagnosis and requiring the services of a genetic counselor. However, because those who have the FMR1 full gene mutation present with a complex physical, cognitive, and emotional phenotype [Reiss et al., 1988; Hagerman, 1991; Freund et al., 1993; Hull and Hagerman, 1993; Mazzocco et al., 1993; Rousseau et al., 1994; Sobesky et al., 1992, 1994a,b, 1995], many genetic counselors are not prepared to address the varied issues of the patient and family members. The genetic counselor should be aware of the clinical phenotype associated with the FMR1 gene mutation.

When a patient or family member presents with cognitive and emotional issues, the physician or geneticist often cannot address these issues or assess the needs and concerns that the patient or family member may have due to the current emphasis on cost-effective medical services and the shortened amount of time that the physician is able to spend with the patient or family member. Consequently, the genetic counselor must address these cognitive and emotional issues in addition to the needs and concerns that the patient and family members bring to the clinic.

With this in mind, it is time to ask two questions: (1) Do genetic counselors understand and address the broad spectrum of clinical involvement associated with the patient carrying the FMR1 gene mutation? (2) Are genetic counselors meeting the needs and concerns of patients and their family members? To answer these questions, the genetic counseling process must be reassessed.

Genetic counseling has been defined as a communicative process that deals with the human problems associated with the occurrence, or the risk of occurrence, of a genetic disorder in the family [American Society of Human Genetics, 1975]. Over the past 25 years, the components of the genetic counseling session have evolved to include (1) obtaining the family history,

(2) determining an inheritance pattern, (3) defining risk assessment, (4) identifying family members at risk, (5) arranging appropriate testing, and (6) conveying and interpreting test results. Genetic counselors are taught that counseling should be educational, nondirective, unconditional, and supportive [Leroy, 1993]. However, to recognize better the patient's cognitive and emotional findings and the patient's needs and concerns, the role of the genetic counselor should be expanded to address these specific areas.

The Fragile X Treatment and Research Center in Denver, Colorado, offers an ideal setting to expand the role of the genetic counselor because it is one of the largest centers serving patients with fragile X syndrome and their family members. Believing that the findings associated with the FMR1 pre and full gene mutation necessitate a genetic counseling session that conveys acceptance, empathy, understanding, support, validation, and guidance, a genetic counseling environment is created that leads to genetic and clinical information as well as personal concerns being shared, heard, and understood. Such an environment builds patient and counselor rapport and leads to ongoing patient contact and support. As an additional tool to aid in the identification of the patient's or family member's needs and concerns, the q-methodology using specially constructed q-sort items has been adapted for use in the genetic counseling setting. An introduction and description of the q-methodology, the q-sort items and process, and the preliminary findings from a pilot study using the q-methodology are presented in this paper.

MATERIALS AND METHODS

Q-Methodology

The q-methodology has long been recognized as a research tool, the framework of which emphasizes the role of communication [Livson and Nichols, 1956]. The q-methodology is based upon a family centered approach, facilitates patient and professional collaboration, reproduces needs and concerns consistent with the subject's own experience, and is useful for constructing relatively objective maps of subjective experience allowing the subject to express personal opinions, values, and beliefs [Stephen, 1985]. It is an instrument for the ranking of items through the process of the q-sort. This ranking procedure is stimulated primarily by the work of Cattell [1952] and Stephenson [1953], researchers in the area of clinical and social psychology. This technique is advised when the salience of one item (relative to other items) is of primary concern as a description of a particular characteristic or construct and, in this instance, formal and informal needs and concerns. Basically, the q-sort technique consists of sorting items into categories that are arranged on a Likert-like scale. The individual doing the sorting is required to place each item into a predetermined number of categories according to a fixed distribution or normal curve. The forced-choice q-sort is an interdependent rank-order procedure that allows for a fixed number of tied ranks [Stephen, 1985]. A value is assigned to each item depending on which column that item is placed by the person doing the sorting. Thus, each item can be compared with all other items, as shown in the q-sort curve in Figure 1.

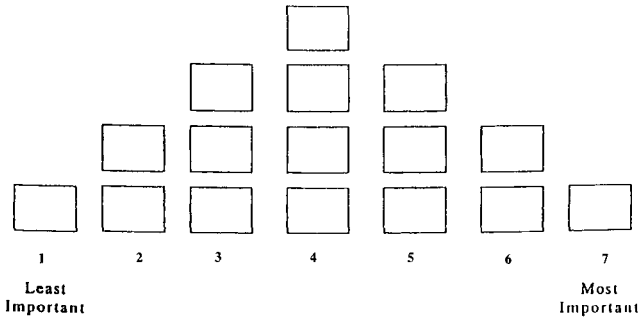

Fig. 1. Q-sort distribution.

Ranking techniques such as the q-sort have notable limitations. There is no way to know how similar in importance two or more particular items are thought to be when these items are placed in the same category or are forced into adjacent categories [Grotelueschen et al., 1976]. For example, the person sorting the cards may consider item A by far the most important goal of all those being ranked. However, items B, C, and D are so close together in the person's mind as to be almost indistinguishable. The q-sort process does not indicate such a relationship. In some instances, two items might be considered equally important but may be forced into adjacent categories, thereby not receiving the same ranking. Another limitation in the q-sort technique is the lack of opportunity for a person to identify items that may not be included in the options offered but which are perceived as important by that person. Therefore, to alleviate this limitation, after completing the sorting process, the subject is asked if there are any items that should have been included in the items constituting the q-sort.

The q-sort has several advantages. In this study, the basis for the use of the q-methodology is the notion that patients being seen for genetic counseling are not having their needs and concerns met. It is believed that patients do have an awareness of their specific needs and concerns, so determining these needs and concerns and assessing the value of the genetic counseling session as it relates to the patient agenda is important. Predetermined items are ranked from "most important" to "least important." Consequently, the subject's score on each item reflects the salience of the item (relative to other items) as a descriptive indicator of the need or concern. In addition, the significance of support items is clearly distinguished in principle from frequency of occurrence. Furthermore, the results of the q-sort are quantifiable and, therefore, subject to statistical description and analysis [Waters et al., 1983]. Response biases and skewing of support items is reduced by requiring items to be sorted into a predetermined number of categories according to a fixed distribution. Finally, the population of participating subjects is diverse, and the q-sort technique provides a way to obtain information that is informative and applicable to the genetic counseling process.

Q-Sort Items

Items in the q-sort were compiled from several sources. One source was a composite of research results over the past 20 years which have defined supportive individuals and behaviors that promote coping strategies in parents of children with special needs. Another source was a study with in-depth interviews with mothers of infants with special needs where results indicated available forms of support and satisfaction with support received [Flynn and McCollum, 1993]. Conclusions from these results guided the formulation of q-sort items.

Other sources that guided the content of support items in the q-sort were several published family support scales. These family support scales included Gottlieb's [1978] Inventory of Socially Supportive Behaviors, Dunst et al.'s [1984] Family Support Scale, and Bailey and Simeonsson's [1988] Family Needs Survey. These social support instruments do not address the specific focus of this study, the identification of the subject's needs and concerns, and the application of these elements to the genetic counseling session. Therefore, items were developed that would directly reflect this focus. The first step in the development of the items was to define and conceptualize the term "needs and concerns" and the subcategories within this definition as they apply to a genetic diagnosis. In the context of this study, needs and concerns were considered a multidimensional construct that includes emotional (grief, sadness, loss of expectations, denial, anger, depression, understanding, and acceptance) and instrumental (material goods and services), and informational (verbal and written details about the child and/or services) functions. These functions serve not only as a mediator of stress, a means to cope with day-to-day or ongoing interactions [House, 1981; Dunst et al., 1986], but also as the primary needs and concerns that a parent brings to the counseling session. The constructs of emotion, instrument, and information are embedded within the items. The categories of formal and informal support are used to delineate the sources of instrumental sources of support for parents [Unger and Powell, 1980]. Thus, the items are divided into formal (any item involving interaction with a professional) and informal (any item involving interaction with a nonprofessional). We determined that 16 items represented the needs and concerns of parents and family members. These items and the categorizations are listed in Table I.

To address the research questions, the q-sort was designed to reflect an equal distribution of support items: 8 items are formal and 8 are informal. The q-sort distribution is reflected in Figure 1, with the 16 support items fitting into the normal curve distribution of the q-sort.

The entire q-sort process takes approximately 15–20 min for the subject to complete, depending on the reading and decision-making speed of the individual sorter. All subjects were literate and capable of completing the q-sort independently.

TABLE I. Rank Order From Highest Noted Need/Concern to Lowest

Item	Category	Median scores
Information on how I can help my child	Formal	6
Special services to help meet my child's needs	Formal	6
Sharing information with others who work with my child	Informal	5
Discussions with medical people about my child's condition or diagnosis	Formal	5
Information about future prospects for my child	Formal	5
Contact with a member of another family who has a child with the same condition or diagnosis	Informal	4
Sharing information with other family members	Informal	4
Financial assistance for expenses	Formal	4
Assistance with child care or respite care	Formal	4
Personal counseling with a professional	Formal	4
Recreation and leisure activities for my child and/or family	Informal	4
Help for my concerns	Informal	4
Support from child's father (or mother)	Informal	3
Participation in an organized parent support group	Informal	3
Discussions of genetic diagnosis, testing and options for future pregnancies	Formal	3
Involvement with a church or strong religious beliefs	Informal	3

Hypotheses

In our study, specific hypotheses were made about fragile X syndrome. Families that have young children with fragile X syndrome versus adolescents or young adults have different needs and concerns. The q-methodology differentiates the needs and concerns among specific age groups. In addition, the length of time that the family has had the diagnosis will result in different needs and concerns between those who have just received the diagnosis and those who have had the diagnosis for a short period of time versus a longer period of time. The q-methodology is also helpful in addressing the different needs and concerns of the father versus the mother of a child with fragile X syndrome.

Fragile X syndrome is unique in that the mother of a child with fragile X syndrome can have a pre or full gene mutation. Research has shown that females with the full mutation have significant emotional problems including "schizotypal features," denial of emotional problems, or other difficulties such as avoidant disorder and panic attacks [Sobesky et al., 1992, 1994a,b, 1995; Freund et al., 1993]. Some females with the premutation have problems with anxiety and mood instability when compared with controls [Franke et al., 1995] and more problems with depression when compared with women with the full gene mutation [Sobesky et al., 1992, 1994b]. Therefore, we hypothesize that females with the

pre and full gene mutation have different needs and concerns. The q-methodology provides a means by which the needs and concerns of females with the pre and full gene mutation can be assessed and compared.

From the q-sort, information can be obtained which will be helpful in guiding the genetic counselor working with family members of those diagnosed with fragile X syndrome. As other centers become familiar with and begin to utilize the q-methodology to guide genetic counseling sessions for members of families with fragile X syndrome, it will become evident that the methodology can be used to identify the needs and concerns of family members who are dealing with other genetic diagnoses.

Subjects

Currently, 37 patients or family members (29 females and 8 males) have participated in the study. Patients and family members serving as subjects for this study had at least 1 child with fragile X syndrome. Subjects were selected according to one of the following criteria: (1) the subject is a female who has been identified through molecular diagnostic testing as a premutation; (2) the subject is a full mutation gene carrier for fragile X syndrome; or (3) the subject is the father of a child diagnosed with fragile X syndrome. Subjects were recruited from families who brought their child(ren) to the Fragile X Treatment and Research Center for a comprehensive evaluation of the needs and interventions related to fragile X syndrome. The subjects had genetic counseling in the past either from a genetic counselor at the center or from a genetic counselor in their geographic area and were scheduled to meet with the genetic counselor at the center during the time of the child's evaluation.

Procedure

At the time of the patient or family member's visit to the Fragile X Treatment and Research Center, the individuals were asked to meet with our genetic counselor. The genetic counselor asked the patient or family member to participate in a research study that was quick, easy, and fun, and one that would allow the center to obtain information about what was most important to them at the time of their visit to the center. The counseling session was defined as a process that had evolved through the guidance of medical professionals and that learning from patients or family members what was important to them was now the emphasis.

With the consent of the subject, a board with 16 rectangular spaces arranged in a distribution curve was placed in front of the subject by the genetic counselor or student volunteer (see Fig. 1 for q-sort distribution). The subject was given 16 small cards upon which were written the items constituting the q-sort.

The subject was asked to read the 16 q-sort items to become familiar with each item. Through the q-sort process, the subject selected and ranked the items according to importance at the time the q-sort was administered. The subject was also asked to turn over any item that was absent or missing from his/her life and whether there was any other item that should have been included in the process. The subject's comments were written down verbatim. The genetic counselor or student volunteer recorded the responses from the q-sort board. A questionnaire designed to elicit socioeconomic information was given to the subject to complete. Confidentiality was maintained by using the last 4 digits of the subject's Social Security number.

Data Analysis

To analyze the data obtained from the q-sort, subjects were categorized into the following subgroups: age of the proband (group 1 = ≤4 years, 9 subjects; group 2 = 5–8 years, 9 subjects; group 3 = 9–12 years, 11 subjects; group 4 = ≥13 years, 8 subjects), length of time that the proband has had the diagnosis (group 1 = <1 year, 13 subjects; group 2 = 1–2 years, 11 subjects; group 3 = ≥3 years, 13 subjects), sex of subject completing the q-sort (29 female subjects vs. 8 male subjects), and mutation status of female subjects (25 with the premutation vs. 4 with the full mutation).

Using the SPSS statistical software package, the data were analyzed by using one-factor analysis of variance. Post hoc tests were used to determine which groups were significantly different from one another. In addition, the mean score of each item was calculated and listed from high score to low score according to the importance of each item to all subjects.

RESULTS

Results obtained from the demographic information revealed that the average age of the proband was 9.5 years, average age of the father of the proband was 38 years, and the average age of the mother of the proband was 37 years. The father's median occupation was classified as executive and the mother's median occupation was classified as professional [Hollingshead, 1957]. The median education of both father and mother was listed as some college. The mean income of all subjects was $35,000–39,000. Six married couples participated in the q-sort.

Information obtained directly from the items (see Table I) that composed the q-sort process revealed that when the age of the proband was examined in relation to each individual item, those who had a child 4 years old or younger diagnosed with fragile X syndrome (group 1) reported the item "personal counseling with a professional" to be absent or missing from their lives more often than did those who had a child aged 9–12 years diagnosed with fragile X syndrome (group 3; $P = .011$).

When the length of diagnosis was examined in relation to each individual item, the item "financial assistance for expenses" was ranked by those who had the diagnosis for 1 year or more (groups 2 and 3) as being more important than those who had the diagnosis for less than 1 year (group 1; $P = .008$). Those who had the diagnosis for 3 years or more (group 3) ranked the item "help for my concerns" as being more important than those who had the diagnosis for 1–2 years ($P = .05$). Those who had the diagnosis for less than 1 year (group 1) ranked the item "information about future prospects for my child" as being absent or missing more often

than did those who had the diagnosis for 3 or more years (group 3; $P = .05$). The item "involvement with church" was ranked by those who had the diagnosis for 2 years or less (groups 1 and 2) as absent or missing more often than did those who had the diagnosis for 3 years or more (group 3; $P = .008$). "Assistance with childcare or respite care" was the item ranked by those who had the diagnosis less than 1 year as being absent or missing more often than did those who had the diagnosis for 1 year or more (groups 2 and 3; $P = .006$).

Male subjects ranked the item "help for my concerns" to be missing more often than did their wives, who were females with the premutation ($P = .012$). The item "support from child's father (or mother)" has never been reported as missing by husbands of females with the premutation.

Females with the full mutation reported the item "support from child's father (or mother)" to be absent or missing from their lives more often than did females who carry the premutation ($P = .05$). Females with the full mutation reported this item missing more often than did those with the premutation and their husbands combined ($P = .05$).

DISCUSSION

Genetic counselors are trained to provide information about the inheritance of a genetic disorder, discuss the manifestations of the disorder, and review treatment options. Details about the availability of diagnostic testing, procedures, and the risks and costs are also often explained [Harper, 1983; Ekwo et al., 1986]. Therefore, genetic counseling is seen as the process of educating and transmitting information. The effectiveness of genetic counseling has been assessed by measuring the ability of the patient to recall the diagnosis and risk probabilities accurately or by the decision to terminate an affected fetus [Kessler, 1989]. However, in actuality, the patient enters the genetic counseling session with multiple needs and a clinical presentation that may impact the counseling process.

The genetic counseling setting is dynamic. The process itself needs to take into account the patient's cognitive and emotional findings and the feelings, motives, anxieties, guilt, anger, denial, needs, and concerns that the patient may bring into the genetic counseling session [Ekwo et al., 1986]. Only by addressing all of these elements during the session can the genetic counselor achieve a rapport with the patient; a good rapport allows the patient to hear the information that is shared and to participate fully in the session, understand the process and, therefore, give the genetic counselor the information needed to cover all aspects of the genetic diagnosis. Such an environment enables the patient to internalize and personalize the information conveyed by the genetic counselor, which is necessary for the patient to make informed decisions regarding medical treatment and interventions, contact other family members if indicated, and communicate with professionals in the community who work with the affected individual [Black, 1980].

The family member of a child who is diagnosed with fragile X syndrome enters the genetic counseling session with feelings and emotions that may change from issue to issue. Such changes require the counselor to be flexible, accepting, empathetic, understanding, supportive, and validating. Often, the genetic counselor working with these patients must normalize but recognize the subtle emotional and cognitive findings associated with the gene mutation and address other complex issues such as the affected child's potential, the state of the parent's relationship, the educational setting for a child with fragile X syndrome, the grief and guilt of the grandparent who carries the gene mutation, pregnancy options, the readjustment of expectations because the affected child is not "normal," the impact of an affected child on other family members, and the consequences of the affected child's life transistions on other family members. When these topics are discussed in the genetic counseling session in a sympathetic and empathetic manner, the patient is able to relax and form a unique relationship with the counselor. The counselor can then offer ongoing support long after the genetic counseling session has taken place, which is an important component of the counseling process for fragile X syndrome because the patient is able to return to the community no longer feeling isolated and alone.

The q-methodology, using the q-sort, offers a unique tool to the genetic counselor for the assessment of the patient's agenda. The q-methodology is the tool by which the genetic counselor can learn, easily and quickly, what is most important and least important to patients and what is missing from their lives at the time the q-sort is administered. From the gathering of this information, one can learn directly from the patient, apply the information to the genetic counseling session, and use the information in offering ongoing support to the patient. In addition, trends from the various subgroups can be recognized and used within the session itself and to educate other genetic professionals about the patient's needs and concerns.

In this pilot study, some preliminary trends have been noted. "Help for my concerns" was ranked as being more important by those who had the diagnosis for more than 3 years. Fathers rated this item as missing from their lives more often than did their spouses who were females with the premutation. Those who fall into these two groups are seldom seen by professionals who can address these concerns. Therefore, this need is one that can be met by the genetic counselor through ongoing contact and support. The item "financial assistance for expenses" was ranked as being significantly important by those who had the diagnosis for 1 year or more, and this finding is an important recognition of the costs of medical and therapeutic intervention related to a genetic diagnosis. The genetic counselor can provide information about financial assistance to the parent(s) of these children. "Assistance with childcare or respite care" was ranked by those who had the diagnosis for less than 1 year as absent or missing from their lives more often than did the comparison groups. Those who had the diagnosis for less than 1 year are often those who have children who are 5 years or younger, and this group finds child and respite care impossible or nearly impossible to obtain. Without child and respite care,

the family unit may suffer insurmountable stress impacting individual relationships within the family. The genetic counselor can recognize this need by addressing with the patient/family member appropriate resources in the community that would provide this service.

Presently, there is no information in the literature that effectively documents what is most and least important to the patient and what is missing from the patient's life. The medical professionals until now have assumed that they know what is important to the patient. However, without patient input, these assumptions should not be accepted. A protocol must be developed, and a reliable research method established that emphasizes the role of communication between subject and professional. The q-methodology as adapted to the medical setting by using the q-sort is such a protocol and research method. Other genetic professionals will be able to use the q-methodology in their clinic settings and so give the patient a voice in the genetic counseling process that up until now has been silent.

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REFERENCES

American Society of Human Genetics (1975): Genetic counseling. Am J Hum Gen 27:240–242.

Bailey D, Simeonsson R (1988): Home-based early intervention. In Odom S, Karnes M (eds): "Early Intervention for Infants and Children With Handicaps: An Empirical Base." Baltimore: Paul H. Brookes.

Black RB (1980): Parents' evaluations of genetic counseling. In PC Health (ed): 3rd quarter. pp 142–146.

Cattell (1952): "Factor Analysis." New York: Harper.

Dunst C, Jenkins V, Trivette C (1984): The family support scale: Reliability and validity. J Indiv Fam Comm Well 1:45–52.

Dunst CJ, Trivette CM, Cross AH (1986): Mediating influences of social support. Am J Mental Deficiency 90:403–417.

Ekwo EE, Kim J-O, Williamson R, Hanson J (1986): Psychosocial aspects of genetic counseling—Factors influencing acceptance of amniocentesis. Grant MCJ-190457; MCH/CCS-86/04, NTIS Order Number PB87-161956.

Flynn L, McCollum J (1993): Support for rural families of hospitalized infants: The parents' perspecitve. Child Health Care 22:19–37.

Franke P, Maier W, Iwers B, Hautzinger M, Froster UG (1995): Fragile X carrier females: Evidence for a distinct psychopathological phenotype? Paper presented at the 7th International Workshop on the Fragile X and X-Linked Mental Retardation. August 2–5, Tromsø, Norway.

Freund LS, Reiss AL, Abrams M (1993): Psychiatric disorders associated with fragile X in the young female. Pediatrics 91:321–329.

Fu Y-H, Kuhl DPA, Pizzuti A, Pieretti M, Sutcliffe JS, Richards S, Verkerk AJMH, Holden JJA, Fenwick RG Jr, Warren ST, Oostra BA, Nelson DL, Caskey CT (1991): Variation of the CGG repeat at the fragile X site results in genetic instability: Resolution of the Sherman paradox. Cell 67:1047–1058.

Gottlieb B (1978): The development and application of a classification scheme of informal helping behaviors. Can J Behav Sci 10: 105–115.

Grotelusechen A, Gooler D, Knox A (1976): "Evaluation in Adult Basic Education: How and Why." Danville, IL: Interstate.

Hagerman RJ (1991): Physical and behavioral phenotype. In Hagerman RJ, Silverman AC (eds): "Fragile X Syndrome: Diagnosis, Treatment and Intervention." Baltimore, MD: Johns Hopkins University Press, pp 3–69.

Harper PS (1983): Genetic counseling and prenatal diagnosis. Br Med Bull 39:302.

Hollingshead AB (1957): "Two Factor Index of Social Position." Mimeograph, Yale University, New Haven, CT.

House J (1981): "Work Stress and Social Support." Reading, MA: Addison-Wesley.

Hull C, Hagerman RJ (1993): A study of the physical, behavioral, and medical phenotype, including anthropometric measures, of females with fragile X syndrome. AJDC 147:1236–1241.

Kessler S (1989): Psychological aspects of genetic counseling: VI. A critical review of the literature dealing with education and reproduction. Am J Med Genet 34:340–353.

LeRoy BS (1993): When theory meets practice. In Bartels DM, LeRoy BS, Caplan AL (eds): "Prescribing Our Future: Ethical Challenges in Genetic Counseling." Hawthorne, NY: Aldine De Gruyter, p 41.

Livson N, Nichols T (1956): Discrimination and reliability in q-sort personality descriptions. J Abnorm Soc Psychol 52:159–165.

Mazzocco MMM, Pennington BF, Hagerman RJ (1993): The neurocognitive phenotype of female carriers of fragile X: Further evidence for specificity. J Dev Behav Pediatr 14:328–335.

Reiss AL, Hagerman RJ, Vinogradov S, Abrams M, King RJ (1988): Psychiatric disability in female carriers of the fragile X chromosome. Arch Gen Psychiatry 45:25–30.

Rousseau F, Heitz D, Tarleton J, MacPherson J, Malmgren H, Dahl N, Barnicost A, Mathew C, Mornet E, Teuada I, Maddalena A, Spiegel R, Schinzel A, Marcos JAG, Schorderet DF, Schaap T, Maccioni L, Russo S, Jacobs PA, Schwartz C, Mandel JL (1994): A multicenter study on genotype–phenotype correlations in fragile X syndrome, using direct diagnosis with probe StB12.3: The first 2253 cases. Am J Hum Genet 55:225–237.

Sobesky WE, Hull CE, Hagerman RJ (1992): The emotional phenotype in mildly affected carriers. In Hagerman RJ, McKenzie P (eds): "International Fragile X Conference Proceedings." Denver, CO: Spectra and the National Fragile X Foundation, pp 99–106.

Sobesky WE, Hull CE, Hagerman RJ (1994a): Symptoms of schizotypal personality disorder in fragile X females. J Am Acad Child Adol Psychiatry 33:247–255.

Sobesky WE, Pennington BF, Porter D, Hull CE, Hagerman RJ (1994b): Emotional and neurocognitive deficits in fragile X. Am J Med Genet 51:378–384.

Sobesky WE, Taylor AK, Pennington BF, Riddle JE, Hagerman RJ (1995): Molecular/clinical correlations in females with fragile X. Paper presented at the 7th International Workshop on the Fragile X and X-Linked Mental Retardation. August 2–5, Tromsø, Norway.

Stephen TD (1985): Q-methodology in communication science: An introduction. Comm Quar 33:193–208.

Stephenson W (1953): "The Study of Behavior: Q-Technique and its Methodology." Chicago, IL: University of Chicago Press.

Unger D, Powell D (1980): Supporting families under stress: The role of social networks. Family Rel 29:566–574.

Verkerk AJ, Pieretti M, Sutcliffe JS, Fu Y-H, Kuhl DP, Pizzuti A, Reiner O, Richards S, Victoria MF, Zhang F, Eussen BE, van Ommen GJ, Blonden LAJ, Riggins GJ, Chastain JL, Kunst CB, Galjaard H, Caskey CT, Nelson DL, Oostra BA, Warren ST (1991): Identification of a gene (FMR-1) containing a CGG repeat coincident with a breakpoint cluster region exhibiting length variation in fragile X syndrome. Cell 65:905–914.

Waters E, Garber J, Gornal M, Vaughn B (1983): Q-sort correlates of visual regard among preschool peers: Validation of a behavioral index of social competence. Dev Psychol 19:550–560.